Dear Sir,

We read the interesting paper of Tsuyama et al. [1] and would like to add our therapeutic experience with a case of Kearns-Sayre syndrome with hyperlactacidemia.

A boy, 9 years and 9 months old, complained of bilateral ptosis, ophthalmoplegia, gait disturbance, atypical chorioretinal degeneration, muscle weakness and hyporeflexia (fig. 1, 2). Blood lactate, pyruvate and serum creatinine phosphokinase were 30 mg/dl (normal 6-16 mg/dl), 0.22 mg/dl (normal 0.3-0.5 mg/dl) and 274 IU/dl (normal up to 1 -0 IU/dl), respectively. A muscle biopsy (nui > -culus quadriceps femoris) showed neither ragged red fibers nor necrosis of fibers. EMU was of the myopathic type. On the basis of the criteria of Rowland et al. [2], a diagnosis of Kearns-Sayre syndrome was made. We assumed an overload of metabolites (lactate) preceding the defect in energy metabolism and a chronic deficiency of end products (coenzyme Q), as a pathogenetic mechanism.

A diet with carbohydrate intake of 7.5 g/ kg/day (minimal dietary allowance), with fat intake of 3.2 g/kg/day (high fat) was administered with the purpose of lowering the high lactate levels. After 3 months, the fasting serum lactate became normal and the ophthalmoplegia, the choreiform movements, the ataxia and the psychomotor disturbances improved.

Thereafter, with an unchanged diet, coenzyme Q10 (‘Juvacor’, Invemdi della Beffa, under licence of Taiyo Pharmaceutical Industry, Tokyo, Japan) therapy was started (30 mg daily) [3]. After 3 months, the fasting lactate was 6.6 mg/dl, the choreiform movements disappeared, bilateral ptosis improved and eye movements were present (fig. 3a-c) with only a mild ophthalmoparesis. This condition persists after 6 months of unmodified diet and therapy. In another personal case of Kearns-Sayre syndrome, no clinical improvement was observed after therapy with coenzyme Q alone. We think that in some cases of Kearns-Sayre syndrome an association of a diet with low-normal carbohydrate, high fat intake and coenzyme Q10 therapy may be useful.

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Fig. 3a-c. Patient after 6 months of therapy (arrows indicate direction of gaze).
References